

Introduction to *Rare Diseases and Scientific Inquiry*

Calling a disease “rare” raises questions. What does it mean to say that a disease is rare? Why should rare diseases be singled out for special attention? In the United States, a disease is considered rare if it affects fewer than 200,000 people. Approximately 7,000 rare diseases are recognized, and researchers continue to describe new ones. Taken together, rare diseases represent a significant health concern affecting over 25 million Americans. Like more-common diseases, rare diseases may be caused by gene mutations, infection from pathogens, and exposure to harmful substances in the environment.

Because rare diseases affect fewer people than common diseases do, they have traditionally been allocated fewer research resources. This has made it more difficult for people with rare diseases to obtain accurate diagnoses of their conditions. Even with an accurate diagnosis, patients may find that there are no existing medications or other treatments to help them. People with rare diseases may feel isolated and even stigmatized. Fortunately, during the past 25 years, increased attention has been devoted to the study of rare diseases, and new treatments are being developed to help patients.

What Are the Objectives of the Supplement?

Rare Diseases and Scientific Inquiry has two main objectives: to help students in grades 6–8 understand

1. that studying rare diseases is not only important to the people affected by the diseases, but it also contributes to understandings that researchers can apply to other, more-common diseases or, more generally, to how the body works and

2. the process of scientific inquiry through studying rare diseases.

The lessons in this supplement help students sharpen their skills in observation, critical thinking, experimental design, and data analysis. They also make connections to other disciplines such as English, mathematics, and social science.

As the supplement achieves its objectives, it helps convey to students the purpose of scientific research. Students experience how science provides evidence that can be used to understand and treat human disease. Ongoing research affects how we understand the world around us and gives us the foundation for improving choices about our personal health and the health of our community.

The lessons in this supplement encourage students to think about the relationships among knowledge, choice, behavior, and human health in this way:

**Knowledge (what is known and not known)
+ Choice = Power**

Power + Behavior = Enhanced Human Health

The final objective of this supplement is to encourage students to think in terms of these relationships now and as they grow older.

Why Teach the Supplement?

Middle school life science classes offer an ideal setting for integrating many areas of student interest. In this supplement, students participate in activities that integrate inquiry, science, human health, mathematics, and

science-technology-society relationships. The real-life context of the supplement's classroom lessons is engaging for students, and they can immediately apply what they learn to their lives.

What's in It for the Teacher?

Rare Diseases and Scientific Inquiry meets many of the needs of teachers in modern classrooms:

- The supplement meets science content, teaching, and assessment standards in the *National Science Education Standards*

(National Research Council (NRC), 1996). It pays particular attention to the standards on scientific inquiry.

- It is integrated with other subjects, drawing most heavily from science, social science, mathematics, and health.
- It has a Web-based technology component that includes interactive activities and simulations.
- Finally, the supplement includes built-in assessment tools, which we note with an assessment icon in each lesson.

Table 1. Correlation of *Rare Diseases and Scientific Inquiry* to Middle School Biology Topics

| Topics | Lesson 1 | Lesson 2 | Lesson 3 | Lesson 4 | Lesson 5 |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------|----------|----------|----------|----------|
| Important levels of biological organization include cells, organs, tissues, organ systems, whole organisms, and ecosystems. | | ✓ | ✓ | ✓ | |
| Specialized cells carry out specialized functions. | ✓ | ✓ | ✓ | ✓ | ✓ |
| Humans have various body systems including those for digestion, reproduction, circulation, excretion, movement, control and coordination, and protection from disease. | ✓ | ✓ | ✓ | ✓ | ✓ |
| Body systems interact with each other. | | | ✓ | ✓ | ✓ |
| Every organism requires a set of instructions for specifying traits. Heredity is the passage of these instructions from one generation to another. | | ✓ | ✓ | ✓ | ✓ |
| Hereditary information is contained in genes. An inherited trait of an individual can be determined by one or by many genes. A single gene can influence more than one trait. | | | ✓ | ✓ | ✓ |
| The characteristics of an organism can be described in terms of a combination of traits. Some traits are inherited, and others result from interactions with the environment. | | | ✓ | ✓ | ✓ |
| Natural environments may contain substances and microbes that are harmful to human beings. | ✓ | ✓ | ✓ | ✓ | ✓ |

In addition, the supplement provides a means for professional development. Teachers can engage in new and different teaching practices like those described in this supplement without completely overhauling their entire program. In *Designing Professional Development for Teachers of Science and Mathematics*, S. Loucks-Horsley and coauthors (1998) write that supplements such as *Rare Diseases and Scientific Inquiry* can “offer a window through which teachers can get a glimpse of what new teaching strategies look like in action.” By experiencing a short-term supplement like this one, teachers can “change how they think about teaching and embrace new approaches that stimulate students to

problem solve, reason, investigate, and construct their own meaning for the content.” The use of supplemental material like *Rare Diseases and Scientific Inquiry* can encourage reflection and discussion and stimulate teachers to improve their practices by focusing on student learning through inquiry.

A correlation of the supplement’s major concepts with the biology and scientific inquiry topics often included in the middle school life science curricula follows (Tables 1 and 2). We hope this information helps teachers make decisions about incorporating this material into the curriculum.

Table 2. Correlation of *Rare Diseases and Scientific Inquiry* to Middle School Scientific Inquiry Topics

| Topics | Lesson 1 | Lesson 2 | Lesson 3 | Lesson 4 | Lesson 5 |
|-----------------------------------------------------------------------------------------|----------|----------|----------|----------|----------|
| Testable questions can be answered through scientific investigations. | | ✓ | ✓ | ✓ | ✓ |
| Scientific investigations use appropriate tools to gather, analyze, and interpret data. | | ✓ | ✓ | ✓ | ✓ |
| Evidence is used to develop explanations and make predictions. | | ✓ | ✓ | ✓ | ✓ |
| Critical thinking is used to relate evidence to explanations. | ✓ | ✓ | ✓ | ✓ | ✓ |
| Alternative explanations are recognized and analyzed. | ✓ | ✓ | ✓ | ✓ | ✓ |
| Mathematics is important to scientific inquiry. | | ✓ | ✓ | ✓ | ✓ |

